quantities to be considered as a possible etiological factor in the causation of compressed air illness.

2. The direct cause of compressed air illness is the presence of air bubbles in the circulation; these gas bubbles consist of nitrogen, carbon dioxide, and oxygen, of which nitrogen is in excess, for the reason that this gas is the chief constituent of the atmosphere, and also does not combine with any of the body tissues.

3. New workers are most susceptible to this disease until the body accommodates itself to this entirely abnormal condition of high air pressure; and, therefore, it is of the utmost importance that only young, healthy men should be allowed to do this work. New workers should, when feasible, be started in low pressures, from which in the course of four or five days they may graduate to become, as they are called, "high pressure men."

MYATONIA CONGENITA, OF OPPENHEIM.

OR CONGENITAL ATONIC PSEUDOPARALYSIS.

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The condition described as myatonia congenita by Oppenheim, of Berlin, and sometimes called Oppenheim's disease, seems to have found but little recognition in America, as no mention is made of it in recent text-books, be they of neurology, pediatrics, or internal medicine. And yet the first autopsy case was published by an American, Spiller, of Philadelphia, and since then a considerable European literature has sprung up around the subject. Possibly one reason for this apparent neglect is the similarity of Oppenheim's appellation to that of myotonia congenita, or Thomsen's disease, which appears invariably to be thought of when the name is mentioned. I may add that there is yet another condition unfortunately similarly named, "Myotonie der Neugeborenen und Säuglinge," of Hochsinger.[1] To avoid confusion, therefore, I have applied the name congenital hypotonic or atonic pseudoparalysis to the disease; this is merely a descriptive cognomen, the pathology being still unsettled.

Before proceeding farther, I shall give the history of three cases (from Oppenheim's Polyclinic at Berlin); the third[2] is somewhat exceptional, and is here published for the first time; the first and second are typical and have been already reported by me.[3]

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[1] Krämpfe bei Kindern, Die Deutsche Klinik, vol. vii.

[2] For permission to publish these case reports I am indebted to Professor Oppenheim.

Case I.—Emma R., aged three and one-half years. The mother brought the child to the clinic because it could not use its legs since birth. The upper extremities have always been normal. The child is, in regard to its intelligence, normally developed. Pregnancy and parturition were nowise complicated. Labor was not prolonged. There was no cyanosis at birth, nor any injury nor acute disease since. Examination shows complete hypotonicity of the lower extremities. Neither patellar nor Achilles reflexes can be elicited. The child can flex the foot upon the leg, but only with much reduced strength. The upper extremities, though they can be voluntarily moved in all directions, also show some weakness. Only if supported on both sides can the child propel itself forward, and this with much effort. Because of the child's excitement, the electrical examination was not complete; but this much was ascertained, that the cruralis and obturator on the left side gave no reaction at all, while the peroneus showed decreased electric irritability.

Case II.—W. J., aged four months. This patient was referred to us from the clinic of Professor Hoffa. The child was born at the normal time; the parturition was easy. There is no history of any trauma. The mother felt fœtal movements until just shortly before labor. The child was at the breast for five weeks, and was then artificially fed. No gastro-intestinal disturbances have occurred. Looks well nourished; the size is that of a normal infant of this age. With the exception of the fingers and toes, the child may have been paralyzed since birth. It moans while being examined and lies motionless. The skin on the legs appears in places slightly mottled and cold to the touch. The patellar reflexes are absent. Complete atony is present. The extremities, being placed in most uncomfortable positions, are left so. Even when irritated by pain (pinch or pin prick) no movement is made, or only very slight ones in the feet and toes, and very gradually. In the arms a similar atony exists. The forearm is bent, the hand pronated, and the fingers held together (slight secondary contraction). Only in the fingers does one see some slow active movements. Even upon pricking with the needle no movement is made. With inspiration the diaphragm is drawn well down, the ribs to which it is attached being thereby somewhat drawn in. The abdomen is distended, the umbilicus pouting. The abdominal reflex is absent. The child cannot as yet sit up. It can move its head. The movements of the eyes are normal. The large fontanelle is closed. The neck is short, but apparently there is no pathological condition here. The cervical spine shows lordosis and lies deep. The dorsal spine is somewhat kyphoscoliotic (doubtlessly congenital). Needle pricks are normally felt in the face. The electrical reaction shows a quantitative but no qualitative change in the muscles of the thighs. In using a strong galvanic current the muscles contract with rapidity.

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[3] Zur Differentialdiagnose der Poliomyelitis anterior acuta, Karger, Berlin, 1908.

CASE III.—A. A., aged one and one-half years, female. There is nothing of hereditary significance. The mother is said to have felt strong fœtal movements. Labor was normal at term, there was no instrumentation, and the child showed no asphyxia. In the fifth or sixth month the mother noticed that the child could not hold its head up, nor sit up (which it had already attempted). Then gradually the present condition developed. On further questioning it appears that the child never really kicked nor used its feet as the normal infant does. It therefore becomes doubtful whether or not the condition was entirely congenital or acquired. At any rate, it seems that an exacerbation occurred in the fifth or sixth month. The child looks well nourished, and has a healthy color. It appears intelligent, notices all that is being said or done. At present it cannot stand, or sit, or hold its head up. In the beginning it sucked well; this has become difficult. There is severe constipation. The head is normally shaped. There is no craniotabes; the fontanelle is closed. The child repeats single words normally. Hearing is good. The pupils react promptly; opthalmoscopically normal. There is a tendency to convergent strabismus. Movements of the facial muscles are normal. Breathing is somewhat snoring. When let go, the child just doubles up as if made of rubber. The head is balanced with much difficulty, and the slightest movement sends it over. Both upper extremities are flabby, lacking all tone, though some movements are to be seen here. The child should be able to raise the arms somewhat in the shoulder. At present, when raised, both limbs again fall back upon the table. In the hands and fingers constant movements are seen, which at first appear athetoid in character, but are certainly more in the nature of play impulses. No reflexes can be obtained. The legs are absolutely atonic. The patellar and Achilles reflexes are absent. A real plantar reflex is not obtainable, only an occasional contraction or extension if this region is irritated. The toe reflex (Bechterew-Mendel) on the left side is mostly dorsal; on the right also, but not constant. The limbs lie extended in extreme outward rotation. Passively lifted they fall back as if lifeless. Active movements are to be seen in the toes. If placed in a very awkward and uncomfortable position, the legs are brought out of this only slowly, and chiefly by movements of the feet. Even when stuck with a needle no lively movement or reflex can be obtained. Occasionally one notices a contraction of the quadriceps. The abdomen is distended. There is no abdominal reflex. The muscles are hypotonic. The diaphragm appears to act normally. The atony of the muscles of the back is especially intense, but so also is that of the hip- and knee-joints. There is considerable adiposis, which masks the atrophy that may be present. There is divergence of the lower part of the thorax. Sensation appears to be normal. The thyroid cannot be distinctly palpated. The percussion note over the sternum is strikingly short. Electrical examination shows marked lessening of irritability for both currents in the lower extremities, less so in the upper. The facial muscles react normally. There is no reaction of degeneration anywhere.

Studying these cases, we find certain points in common—a rather extensive apparently flaccid paralysis, with absence of patellar and Achilles reflexes. In one there is vasomotor disturbance, in another slight atrophy, masked, however, by adiposis. Viewed thus far these cases might be taken for anteropoliomyelitis. A little closer analysis, however, will bring out certain interesting facts—namely, that Cases I and II with certainty, and Case III quite probably, date from birth, or, in other words, are congenital. Now, congenital cases of anteropoliomyelitis appear not to exist. The one case cited in the literature[4] belongs possibly to the group of cases I mean to describe[5] and has nothing to do with poliomyelitis, as two other children of the family were similarly afflicted (familial intra-uterine poliomyelitis!), and the paralysis was progressive—increased instead of decreasing, as is the rule in anteropoliomyelitis. Furthermore, there is no really degenerative atrophy present with reactions of degeneration. The paralysis which is extensive, especially in Cases II and III, has not gradually confined itself to an area smaller in extent than that which was at first affected, as is almost invariably the case in anteropoliomyelitis. Finally, on careful examination, one sees that some movements are present in the affected muscles, that we are not really dealing with genuine paralysis, but with an extreme condition of muscular atony. These muscles feel soft and doughy, and, excepting in the one case, not cold to the touch. We have, therefore, before us a symptom complex which deviates widely from that found in infantile spinal paralysis.

In 1900 Oppenheim[6] described a condition of general or localized atony of the muscles, concerning which he said he had found nothing in the literature. These cases occurred within the first two years of childhood, and showed as especially characteristic a hypotony or even atony of the muscles combined with partial or complete loss of the tendon reflexes. The limbs, particularly the lower, were at times so flaccid that they could be shaken like flails. There was in all cases an impairment of active motion. When the condition was extreme, the limbs lay perfectly still, and it appeared as if complete paralysis obtained. But on more careful observation, one could note occasional contractions in various muscles, which, however, were feeble, and lacked sufficient energy to give locomotive effect. Less involved cases showed only hypotony and a marked weakness. In one case the muscles of the trunk and neck were affected, besides those of the limbs, so that the infant of eight months could neither sit up nor support its head. Even in so generalized a condition, however, the muscles of the eyes, tongue, and throat were unaffected, as also the diaphragm. The intercostals, on the other hand, appeared to be involved. The muscle felt flaccid and soft, and appeared to be thin, though one could scarcely apply the term atrophic. Electrical examination showed in the severer cases a considerable quantitative, though never qualitative, alteration, up to even complete disappearance of reaction. In mild cases the reaction was normal. In but one case tardy contraction, and this only in portions of the muscle, was observed. No abnormalities, so far as it is possible to judge in such young infants, were discoverable in the sphere of intelligence, sensory system, or special senses. The malady always appeared to be congenital. In its typical form it seems to be the antithesis of Little's disease.

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[4] A. Sevestre, Ref. Neurol. Zentral., November 9, 1900, S. 904 (Paralysie flasque les quatre membres et des muscles du tronc, etc., Bull. de la Société de pédiatria de Paris, Fév.-Mars, 1899).

[5] H. Oppenheim, Lehrbuch, p. 224.

[6] Ueber allgemeine und lokalisierte Atonie der Musculatur (Myatonia) im frühem Kindesalter, Monatsschrift f. Psychiatrie u. Neurologie, 1900, Band viii, Heft 3, p. 232. Ueber einen Fall von Myatonia congenita, Berl. klin. Woch., March 7, 1904, No. 10, p. 255; also Berliner med. Gesellschaft, Sitzung, February 24, 1904.

Oppenheim thought the pathological factor of this condition was to be sought, not in the central nervous, but in the muscular system, which was probably retarded in its development—for not only was he able now and again to observe partial or complete return to the normal within the course of time, but because he never had occasion to notice this condition in older children. He did not think it had aught in common with progressive muscular dystrophy. He did not entirely exclude the possibility of disease in the anterior horn cells prompting the developmental inhibition of the muscles.

Shortly after other cases began to appear in the literature, two of Berti's,[7] and then one, the first with necropsy, of W. G. Spiller's.[8] This last case was a child of twenty-two months. The head, trunk, and extremities were affected. There was very great hypotonicity of all the limbs, especially the lower (which could be flexed so as to bring the thighs in close contact with the trunk, and the feet easily behind the head). The child could not stand, sit unaided, hold up its head, or hold anything in its hand. It had always been constipated. The faradic electrical response was retained. Besides convergent squint,[9] certain abnormal conditions of the internal eye obtained, which, as they have nothing to do with the general condition, need not further be considered here. The child died during fever of unknown cause. The autopsy, made twenty-four hours post mortem, showed absence of postmortem rigidity and a hypotonicity of the limbs as great as that during life. The muscles were flabby. Section of the calf showed fat 6 mm. in thickness, with very little muscle, and this paler than normal. There was hardly any muscle in the sole of the left foot. The subcutaneous fat over the triceps measured 3 mm. The brain weighed 980 grams; the dura was adherent. The spinal cord and brain were well developed; the anterior and posterior roots normal. Microscopic examination of the brain, cord, nerves, and muscles (made by Professor Spiller personally) showed normal cells in the anterior horns of the cervical and lumbar regions (thionin stain), and a normal condition of the nerve roots. No spinal meningitis was present. There was no degeneration in the pyramidal tracts. Marchi sections showed no degeneration. The optic nerves were normal (Weigert and Marchi). The nerve cells of the paracentral lobules appeared normal, but in the section Betz's cells were not numerous. Various nerve sections, differently stained, were normal. Muscles from the sole of the foot, back of the trunk, and the left calf had a hyaloid appearance, and those from the sole of the foot were striking because of the large amount of fatty connective tissue, and on account of the considerable increase in the nuclei of this tissue. The muscle fibers were small, those from the sole and the calf much smaller than those from the back of the trunk (the child had more power in the muscles of its back than in those of its lower limbs). The transverse striations were well preserved, the longitudinal not so distinct. Nerve fibers within the muscle from the sole of the foot appeared normal. The mesenteric lymph glands were much enlarged; the organs of the body were macroscopically normal.

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[7] Contribuzione all' Atonia muscolare congenita di Oppenheim (Terza adunanza della sezione Emiliana della Società Italiana di pediatria, 4 Dez. 1904, in Pediatria, February, 1905, p. 134).

[8] General or Localized Hypotonia of the Muscles in Childhood, Univ. Penna. Med. Bull., 1905, xvii, No. 11. See also Bernhardt, Neurologisches Zentral., 1907, No. 1. Spiller, Ebenda, No. 11.

This report strengthens the surmise of an inhibited development of the muscle fibers. Quite different, however, are the findings of Bing,[10] who examined a piece of excised muscle from a living child suffering from typical congenital myatonia. Examination of this muscle tissue showed, apart from probable increase in the nuclei, an absolutely faultless structure.

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[9] This was the "ordinary concomitant convergent squint, and not an internal strabismus due to paralysis of the external rectus." (G. E. de Schweinitz, cited by Spiller.)

[10] Ueber atonische Zustände der kindlichen Muskulatur, Med. Klinik, 1907, No. 1.

A report of the microscopic examination of the internal organs in Spiller's case was published by A. J. Smith.[11] The heart, pancreas, kidneys, liver, and bladder were normal. The thymus showed distinct intralobular sclerotic changes (fibrosis), with enlarged Hassal's corpuscles, and arteriosclerosis (thickening of the adventitia, occasional fibrosis of the intima, and proliferation of the endothelial cells). The spleen also showed sclerotic changes in various bloodvessels and endothelial proliferation of the Malpighian bodies. The mesenteric glands showed proliferation of the lymphatic cells.

A little patient whom I had occasion to examine with Stabsarzt Helmer, on the pediatric division of the Königlische Charité in Berlin, last year, also had this affliction. The diagnosis had been made by Dr. Reyher, who had sent the child in from the polyclinic, and who later, the child dying of pneumonia, examined some of the muscles microscopically.[12] His report to me was as follows: Side by side with patches of normal muscle fibers were extremely altered ones. The abnormality consisted in the cross-striation being indistinct or entirely absent. In some parts the longitudinal striation appeared to be lost. The individual muscle fibers varied very much in breadth, some being abnormally thin, others apparently abnormally broad. Where the changes were most pronounced there were but few muscle fibers to be found at all, fat and connective tissue rich in nuclei being seen instead. The changes were most marked where the clinical symptoms had been most manifest, as in the lower extremities. The long bones of the lower extremities showed considerable atrophy (which the Röntgen photogram had already shown in vivo). Then came the reports of a whole series of new cases, among which was one with necropsy.[13]

The patient was aged four months, of healthy parents (who also had another perfectly healthy four-year-old child). Parturition was normal. The infant never had had convulsions. From birth on, the neck, trunk, and all extremities appeared to be completely inert. The condition neither improved nor progressed. Examination showed a generalized flaccid paralytic condition, the muscles of bulbar innervation alone being exempt. At times a slight internal strabismus of the right eye was noticed. The facial muscles were normal. The infant sucked and swallowed well. Its cry seemed weaker than that of infants of like age. The muscles of the neck could not support the head. Those of the trunk lacked

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[11] Histological Changes Encountered in the Thymus and Elsewhere in a Case of Congenital Hypotonia, Univ. Penna. Med. Bull., October, 1905.

[12] This case was to be published in full.

[13] A. Baudouin, La myatonie congénitale (maladie d'Oppenheim), Semaine médicale, May, 1907, No. 21.

all tonicity. The intercostals were also involved. Only in the fingers and toes did one notice movements, and these were very feeble. The feet showed varus, with slight equinus. All reflexes except those of the eye were absent. The skin, especially of the lower extremities, was wax-like and glossy, and showed a swelling resembling myxœdema. No other atrophic disturbances were present. Sensation, the sphincters, and the special senses were normal. There were no reactions of degeneration anywhere. The bony system showed neither signs of rickets nor syphilis. The child died of a bronchopneumonia, having had convulsions one hour before death. The autopsy was made twenty-four hours post mortem. The external aspect presented nothing of special interest. The muscles were pale and sunk in fat. There was considerable subcutaneous fat, in places 1 cm. thick. Microscopic examination of the nervous system showed much of interest. Sections from the convolutions of Rolando stained with the Nissl method were found normal. The nucleus of the third nerve (of both sides) was normal. That of the sixth and twelfth nerves was formed of cells well developed, many of which showed chromatolysis. Stained with the Pal and Marchi methods, the cord showed nothing abnormal. A section from the lower region of the eighth cervical segment stained with the Nissl method showed the cells of the anterior horns to be diminished in size and less numerous than normal. Those of the column of Clarke were well developed. The vessels were normal. No neuronophagia was observed. A similar condition was found in other parts of the cord. Baudouin concluded that there had been an arrest in the development of the anterior horns without any inflammatory phenomena.

A section of the anterior and posterior roots of the third lumbar segment showed a considerable difference between the thickness of the two, Normally the anterior is about one-half the size of the posterior. Here it was one-quarter to one-fifth the size. The posterior also stained much darker, the bundles of fibers having less amorphous tissue between them.

The sciatic, as an example of a peripheral nerve, showed no abnormalities by the Marchi method (no regressive or degenerative lesion therefor). The Weigert and hematein-eosin stain made manifest, however, that there were axone cylinders in the nerve not yet myelinated. There was also a slight sclerosis, and the nuclei of the sheath of Schwann were slightly more numerous than usual. The same condition was found in the crural, median, and cubital. Stained with osmic acid the right internal motor oculi is normal; the external, however, shows an absence of many myelin sheaths (the infant displayed occasional internal strabismus of the right eye). Here, too, Baudouin concludes there is an arrest in development and no neuritis. Finally, the muscle showed great changes. With weak magnification the predominating picture was that of intense sclerosis. The stands of fibrous connective tissue within the muscle were numerous and thick. The vessels were also a little thickened, but especially interesting was the fact that they were surrounded by a zone of young cells which stained deeply, among which one noticed a rather large number of eosinophile myelocytes. As in the Charité case, one saw here bundles of very attenuated muscle fibers (measuring 6 to 8 microns) alongside of very thick hypertrophied ones (measuring 100 microns in diameter). The nuclei of the sarcoplasma were increased in number, and sometimes invaded the fiber itself. Longitudinal sections showed a disappearance of the transverse striation, the longitudinal being clear. The same picture was seen in the various muscles examined, the diaphragm, however, being the least affected. These lesions were, in Baudouin's opinion, of a regressive nature, such as one sees in the myopathies. Hystologically the skin was normal save that the subcutaneous fat was very abundant. The kidney, suprarenals, liver, and pancreas were normal. In the spleen there was a mild sclerosis. Some of the Malpighian follicles were a little more evident than normal. The thyroid presented intense sclerosis involving the acini, these being filled with cells and containing no colloid substance. No lymphatics could be seen. The thymus showed a similar sclerosis.

Finally, the gross and microscopic findings in the case of W. J. (Case III of my series) were reported by M. Rothmann[14] (in whose clinic the infant came to autopsy[15]). There was an absence of rigor mortis; the muscles all looked pale and thin. The subcutaneous fat was strongly developed. All segments of the cord showed extensive disappearance of the anterior ganglion cells, with increase in the capillaries, and atrophy of the anterior roots. In the hypoglossus nuclei there was beginning disappearance of the ganglion cells, with chromatolysis. The muscles showed atrophy and small-celled infiltration. Another case with necropsy was announced by MM. Variot and Devillers[16] at the Société de pédiatrie, October 15, 1907. Only a partial autopsy, it seems, was made; the results of the histological examination are still to be published.

Besides the case here communicated for the first time, some 34 or 35[17] cases have thus far been reported (Oppenheim 5, Berti 2,

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[14] Dr. Rothmann had the kindness to write me, at the time the autopsy was made, that the macroscopic examination proved to be negative.

[15] Berl. med. Woch., November 12, 1908; Jahresver. d. Gesell. Deut. Nervenärzte, Heidel., 1908.

[16] Société de pédiatrie, October 15, 1907 (Archives de médecine des enfants, November, 1907, No. 11, p. 700).

[17] A case of Beevor's ([33]) is cited by Collier and Wilson under their bibliography. I have not, however, been able to locate it. The case of Schüller ([37]) was but poorly reported, and the diagnosis, therefore, indefinite. The same may be said of a case reported by Hutinel ([64]).

Muggia,[18] Spiller, Kund,[19] Rosenberg,[20] Jovane,[21] each 1, Sorgente,[22] Comby,[23] each 2, Tobler,[24] Bing, Leclerc,[25] Baudouin,[26] Misserey,[27] Cattaneo,[28] Variot,[29] Reyher and Helmholz,[30] each 1, Haberman 2, Lugenbühl,[31] Beevor,[32] Thomson,[33] each 1, Batten[34] 3, Collier and Wilson[35] 4). Three cases (2 by Bernhardt[36] and 1 by Schüller[37]) have been published under this heading, but do not in all probability belong here, while 1 (Carey Coombs[38]), though classed among the myatonic cases by Collier and Wilson, appears to me to be far too atypical to be accounted such. Another case (Wimmer[39]) resembles congenital hypotonic pseudoparalysis only in part, and is to be classed rather with the progressive spinal atrophies.

Studying the reports of these cases (and excepting for the time being those published in England—of which I shall presently speak) one may conclude the following: Neither sex predominates. Heredity was nil. Pregnancy and parturition were normal. In several cases fœtal movements were not felt, in others they were felt precisely as in former pregnancies. In most cases nutrition was very good. In many adiposis was noticed. These children were somewhat pale and flabby, growth was normal (in no case were symptoms of rickets noticed[40]). The muscles were soft and doughy, but were never reported as having been truly atrophic. In one case the lower extremities felt cool and looked mottled;

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[18] Un caso di paralisi completa congenita dei quattro arti (1° reunione dei pediatrie Piemontesi, 12 Fev., 1903, in Pediatria, Mars, 1903, 179).

[19] Ueber Myatonia congenita (Oppenheim), Diss., Leipzig, 1905.

[20] Ueber Myatonia congenita (Oppenheim), Deutsch. Zeit, f. Nervenheilk, Band 31, 1906.

[21] Contributo clinico allo studio dell' atonia muscolaro congenita di Oppenheim, La Pediatria, Mars, 1906, xiv, No. 3.

[22] Due casi di atonia muscolare congenita di Oppenheim, La Pediatria, 1906, No. 5, xiv.

[23] Atonie musculaire congénitale in Archiv. de Méd. des Enfants, 1906, Band ix, No. 9, 552; Société de Pédiatrie, October 15, 1907 (Archives de médecine des enfants, November, 1907, No. 11, 700).

[24] Ueber Kongenitale Muskelatonie Jahrbuch f. Kinderheilkunde, 1907, Band 66, Heft 1.

[25] Un nouveau cas d'atonie musculaire congénitale (Maladie d'Oppenheim), Gazette des Hôpitaux, December 10, 1907, No. 141.

[26] Loc. cit.

[27] Myatonie Congénitale ou Maladie d'Oppenheim, Thèse, Lyon, 1908. (Same case as Leclerc, No. 25.)

[28] Sulle paralisi dei neonati e sulla myatonia generalizzata di Oppenheim (Clinica moderna, June 13, 1906, 282).

[29] Loc cit.

[30] Provisional report at the Breslau Congress for Pediatrics, 1908.

[31] Ein Fall von Myatonia congenita, Deut. med. Woch., August 29, 1907, 1430.

[32] A Case of Amyotonia Congenita, Brain, 1907.

[33] A Case of Amyotonia Congenita, Brain, May, 1908, 160.

[34] Three Cases of Myopathy, Infantile Type, Brain, 1903, 147.

[35] Amyotonia Congenita, Brain, May, 1908, xxx.

[36] Zur Kenntniss der sogenannten angeborenen muskelshlaffheit, Neurol. Centralblatt, 1907, Heft 1.

[37] Wien. klin. Wochenschr., June, 1904, No. 25, 722 (Verien f. Psych. u. Neurol. in Wien, May 10, 1904).

[38] Congenital Hypotonia (Congenital Amyoplasia), Brit. Med. Jour., June 15, 1907, 1414.

[39] Archiv f. Psych und Nerv., No. 42, 1907.

[40] Rosenberg's case "should have had a light attack of rickets in the eleventh month," no symptoms, however, being present at the time of examination.

in four a myxœdematous condition of the skin was described. Otherwise no trophic disturbances were noticed. The cranial nerves were always normal (in Spiller's case congenital amaurosis obtained, probably of central origin, the optic nerve being found normal). Sphincters, sensory system, and intelligence were normal. In all the patellar reflex was absent, in some the skin reflexes also. The electrical response was very much diminished or entirely absent, but never qualitatively altered. In all cases the chief symptom was the immobility of the limbs—the inactivity of the child. It appears most probable that in all cases the condition dated from birth (the only doubtful cases being Case III, which I report, and which Professor Oppenheim also thought more probably congenital, and Rosenberg's, in which the atony was first noticed in the eleventh month—the mother, however, having scarcely felt any fœtal movements at all), and was in most cases noticed within the first few days after birth. In several it was only discovered later, when the first attempts at walking were made. In most cases it was found (in others the fact is not reported) that movements or "remnants" of movements were present even in the atonic muscles, if not active, then at least on pain-irritation. In a few, absolute immobility seemed to obtain (in Muggia's case in all four extremities, in Jovane's and Sorgentè's cases only in the lower), and here no improvement occurred. In other cases in which at first no movement was seen this later became manifest.[41] One even noticed that the patellar reflex, and also single active movements set in temporarily after energetic faradization.

Many cases (12) showed improvement after long-continued treatment (Rosenberg reports improvement even after thirty-one months). In others no improvement occurred. In several cases the outcome is not known. In our third case an exacerbation was noticed in the fifth to sixth month. In no other case was the condition noticed to progress. In 11, death occurred immediately, due to pulmonary disease (pneumonia). Three cases had convulsions. This occurred in Sorgente's 2 cases (both of the same parents), in the one, tonic and clonic spasms of all the limbs five hours before death; in the other, generalized convulsions twice in the last twenty-four hours; in Baudouin's case, one hour before death. Sorgente's cases suggest that the condition may occur familialy.

In all the cases the lower extremities were affected; in more than half the upper also (though usually in lesser degree); in many the trunk besides; in five, also the head; in two, the diaphragm seemed affected; in three, strabismus was present; in one, the facial muscle appeared somewhat weak. Throat and cranial nerves, therefore, appear on the whole to be free. Never when movements were executed was coördination disturbed. Standing and walking were in no case possible. Most all showed a functional kyphosis usually very intense in sitting. In two, atrophy of the bones of the lower limbs was noticed. Once, secondary contraction (in the hand). The distal parts were always less affected than the proximal, and were even entirely free in several cases of severe proximal involvement. Slight cases showed hypotonicity only; severe cases absolute atony. In most cases the teeth showed no abnormality; in one or two, teething was late. Subjective symptoms do not appear to occur.

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[41] It may here be remarked that in some of the histories little attention was paid to these slight movements in the muscle groups, or the fact was not reported. In all cases which were carefully examined and minutely reported the fact is emphasized that nowhere did absolute immobility obtain, so that we may conclude that actual paralysis occurred in no case, only varying degrees of atony.

The majority of the cases reported in England (Batten, Collier and Wilson, Coombs, Thomson) are of older children (five to ten years of age). In most of these contractures were noticed (doubtlessly due to the long continuance of the disease). The affected muscles are defined as being small and thin, and the description suggests considerable wasting, even though no real atrophy should have occurred. In no case was adiposis present (this period of childhood being less prone to adiposis than the first two years). These authors emphasize the long, narrow, pad-like formation of the feet and hands. The joints are especially hyperextensive. Certain of these cases show discrepancies which make them somewhat doubtful. Thomson's evidently had a difficult partus (under chloroform). The mother complained of the child being drowsy. The hamstrings and adductors of the thigh did not appear to be affected (distribution?). There was little difference between CCC and ACC. Finally, the patellar reflexes were present, and the improvement was so rapid that the child could sit up and make attempts to walk after four months. One case of Collier and Wilson's was well up to the twelfth month. These authors speak of a prenatal type, and even assume the occurrence of this disease after acute illness in a previously healthy child. The major evidence as yet in hand is, I believe, against this inference. Coombs' case is certainly too atypical to belong here. The child was ten years old. The condition was first noticed in one hand, and only months after in the rest of the body. The distal muscles suffered more than the proximal. The scapulæ were winged. The knee-jerks were present and brisk. The Achilles phenomenon was also present.

The clinical picture of myatonia congenita is, on the whole, therefore, a typical one, and may roughly be landmarked as a congenital, atonic pseudoparalysis, met with in children in the first two or three years of childhood, in which the reflexes are very weak or abolished, electrical reaction reduced but never degenerative, real atrophy not met with, the distribution proximal, with no progression, the muscles of bulbar innervation practically exempt, and no sensory, bladder, nor rectal involvement occurring. In a minimal group the children are older, contractures are present, the muscles small and thin (atrophic?).

Coming to the differential diagnosis of this disease, we must first consider those conditions in which we often meet with a high degree of muscular atony—namely, infantile or congenital myxœdema, mongolianism, and, above all, rickets. Among our cases we have, in fact, four (cases of Baudouin, Leclerc, Berti) which showed myxœdematous thickening of the skin: while in one (Baudouin's) the autopsy displayed gross changes in the thyroid gland. And yet I believe we may exclude myxœdema entirely. In the first place, this symptom complex is typical: the characteristic physiognomy; the small, widely separated eyes; the spare hair growth; the swelling about the face, forehead, and especially the eyes (regions not at all affected in three of the cases above referred to); the pallor or wax-yellow sallowness of the skin, and it's great dryness (often scaling); the thick tongue, usually protruding from the mouth; the umbilical hernia, obtaining in all cases of any severity; the persistent patency of the large fontanelle; the impaired psychic condition; the pronounced disproportion between length and breadth of the body; and finally, the fact that myxœdema is unusual in boys, and that only one of the myatonia cases observed (Baudouin's) bears the slightest resemblance to it. On the other hand, no condition described as arising actually or experimentally from thyroid insufficiency[42] has any likeness to Oppenheim's disease.

As regards mongoloid idiocy, it may be dismissed without comment, as it gives so typical a picture that it could scarcely be mistaken for anything else.

Rachitis, however, we must study more carefully, as this disease often evidences a high degree of hypotonicity, which may easily enough be taken for paralysis. Rickety limbs may occasionally be placed in all sorts of abnormal and uncomfortable positions without causing distress to the young patients. They may in some cases even be twisted about after the fashion of the circus "snake-man," strongly hyperextended and shown to be loose and wabbly like flails. This pseudoparalytic condition occurring in rickets has been described by Oppenheim,[43] Comby,[44] Vierordt,[45] Hagenbach-Burckhardt,[46] and Bing.[47] One may also find a slight muscular atrophy in these cases not easily noticed because of the frequent adiposis present. Whether such fully developed condition of rickets can be present at birth is highly questionable, and has up to recently been the cause of much controversy. Escher,[48] who undertook the microscopic examination of sections from a large number of apparently rachitic newborn infants, could at no time find any changes which would substantiate the diagnosis of rickets. Heubner[49] holds that one will rarely if ever be able to make the diagnosis of congenital rickets if one believes with Pommer (with whom he likewise agrees) that a retarded bone development, a standing still in the half-completed stage, is the cardinal feature in rickets. And Stoeltzner[50] says that no unchallengeable case of congenital rickets has yet been recorded. Most conclusive, however, has been that painstaking study of nearly a thousand newborn infants by E. Wieland,[51] of Basel, undertaken, as he tells us, to prove the positive side of this question, but which led to an absolute negation. He shows how former mistakes were made in judging the costocartilage nodes (Kantenbildung) so often met with in the newborn, to be rachitic ("rosary"). These are normal occurrences, especially pronounced in strong muscular babies (so also Feyerabend, Cohn, Spietschka, Escher, and Tschistowitsch). Another symptom which led astray was the so-called "craniotabes." This symptom he found present in 18 per cent. of his material (that is, in 182 of the 1000 cases), a number of these also showing bone defects, the infants being otherwise absolutely normal and not developing any signs of rickets (these cases being followed up from one to two years). He believes that the condition of "soft skull" is a "pathology" sui generis, occurring in weak children, and having no connection with rickets. Wieland's analysis leads us to conclude that rickets cannot be clinically demonstrated in the newborn, and that in all probability not the disease itself but only the disposition to it may be congenital. We may therefore say that in the majority of cases of myatonia congenita the condition is noticed within the first few days or weeks post partum, this never occurring in rickets. In most of the cases of rachitic pseudoparalysis observed by Vierordt[52] the children had already learned to stand or walk, only again to "unlearn" it. The rachitic child does not make attempts at walking, but, contrary to the normal infant, draws up its legs and usually shows resentment (irritability) to passive motion. In myatonia no rickety symptoms have thus far been reported. And finally, this fact of interest may be noted, that the study of rickets undertaken in Hagenback's clinic at Basel showed that the tendon reflexes in this disease are never absent, but even exaggerated at times, while in myatonia congenita they are always absent.[53]

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[42] Ewald in Nothnagel's System, Band xxii; Kassowitz, Infantile Myxœdem, etc. Vienna, 1902; Magnus-Levy, Organotherapie und Innere Secretion, 1906; Krehl, Ueber die Störung chemischer Korrelationen im Organismus, 1907; Anton, Entwicklungsstörungen beim Kinde, Berlin, 1908.

[43] Lehrbuch, second edition.

[44] Grancher's Traité des maladies de l'enfance, tome iv.

[45] Ueber Hemmungslähmungen im frühen Kindesalter, Deutsch. Zeit. f. Nervenh., 1900, Band xviii, S. 167.

[46] Klinische Beobachtungen über die Muskulatur der Rachitischen, Jahrb. f. Kinderheil., 1904, lx, Heft 3.

[47] Loc. cit.

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[48] Escher, Zur Frage der angeborenen Rachitis, Jahrbuch f. Kinderheil., Band lvi, S. 613.

[49] Lehrbuch, Band i, S. 667, 668.

[50] Handbuch d. Kinderheil. of Pfaundler u. Schlossmann, Band i, 605.

[51] Klinische Untersuchungen über Frührachitis, Deut. med. Woch., September 3, No. 36, 1545.

[52] Loc. cit.

But little need be said concerning the "inhibition paralyses" ("Hemmungslähmungen") of Vierordt.[54] These are pseudoparalyses due to pain, occurring in Barlow's disease, in hereditary syphilis (Parrot's disease), and also in rickets, as already described. As they are due to pain, the very examination will call forth muscular spasm (Oppenheim). Besides, it has been pointed out that syphilitic pseudoparalysis very rarely affects the lower extremities.[55] The history of these cases would also show that the paralysis did not date from birth (only one congenital case has been reported[56]), and that symptoms of syphilis are present, or were so a few weeks previously.[57] Infantile scurvy would also give its symptoms (swollen gums, tendency to hemorrhage, etc.), and the history would probably point to error in diet. Only once, to my mind, has a painless pseudoparalysis due to scurvy been mentioned.[58] The disease is never congenital. In myatonia congenita no symptoms of scurvy or syphilis have been noticed, no pain seems to have been present, and the affliction has been preëminently in the lower extremities.

As to polyneuritis, only that due to diphtheria could come into question, and in fact only the nasal type, in which the diphtheria might be overlooked and the history give no account of it. Then, too, diphtheritic polyneuritis within the first two years of infancy is a rarity. The muscles here affected would give the reaction of degeneration.

Paralysis due to poliomyelitis, even if not congenital, has been seen to occur early (one case of Duchenne on the twelfth day, one of Zappert on the fifteenth, one of Sinkler in the sixth week), though such early cases are extremely rare. It is easily differentiated from myatonia by the atrophy and trophic disturbance, the reaction of degeneration, the course of the disease, and in the fact that poliomyelitic paralysis occurs after an acute illness in a previously healthy child. This disease is rarely symmetrical, myatonia congenita probably always; and finally, infantile spinal paralysis improves rapidly, only a (small) part of the original paralytic involvement remaining permanent—the improvement in myatonia being very slow, and general, not in any single limb.

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[53] The knee-jerk was reported present in Thomson's case, though nothing is said of its character. This case is not entirely unchallengeable.

[54] Loc. cit.

[55] Hutchinson, Lectures on Diseases of Children, London, 1906.

[56] Vicarelli, Revue Mens., Mars, 1892.

[57] This was noticed in a case of Oberwarth (Zur Kenntnis der syphilitischen pseudoparalysis, Jahrbuch f. Kinderh., 1899, Band xlix). It is also interesting to note that in a few cases of this series no swelling could be felt in the involved limbs, and in four cases there was no pain on pressure or passive motion.

[58] Vierordt, loc. cit.

We may exclude the possibility of a spinal hemorrhage (hematomyelia) entirely, as there are no sensory disturbances, nor affection of the bladder and rectum, nor decubitus. Nor is there anything in the etiology to suggest it (forceps, difficult labor, convulsions, scurvy, infectious disease). Should, however, the possibility of spontaneous hemorrhage in uteri be admitted (such hemorrhages occur in the adult), and we surmise that such hemorrhage has occurred in the anterior horns alone, antepartum, we would expect to find a degenerative paralysis present—in other words, the picture of congenital anteriopoliomyelitis—which, as we have already concluded, does not appear ever to have been seen.

We finally come to the consideration of two conditions which have been associated by one or two writers on this subject with our congenital hypotonic pseudoparalysis, and which, in view of Baudouin's pathological findings, are especially interesting in this connection. The first is a motornuclei aplasia, and the second is the spinal type of muscular atrophy, with which we may also consider the muscular dystrophies.

There is a pathological condition which manifests itself through paresis or paralysis of the muscles of the eyeball (ophthalmoplegia), through atrophy of the tongue, through inadequacy of facial mimic, etc. Sometimes all these symptoms are present, sometimes only one or two, as ophthalmoplegia or ptosis alone. These symptoms correspond to a lesion in the nuclei of the oculomotor, facial, hypoglossal, etc., and were thought by Möbius[59] to be due to an absence or partial development of the bulbar motor nuclei ("Infantiler Kernschwund"). Heubner,[60] in an autopsied case, was able to prove the correctness of this surmise (though it appears that some of these cases may be due to a congenital lesion anywhere along the tract between the motor nuclei and the muscle[61]). That a similar aplasia or hypoplasia of the anterior horn cells in the lumbar or cervical segments might occur does not seem impossible, but no such case substantiated by autopsy or case even suggesting such occurrence have I been able to find in the literature,[62] and the autopsies of Spiller showed no trace of spinal involvement, while that of Baudouin showed that much else apart from the anterior horn cells was pathologically affected. Muggia thought that some type of spinal aplasia might be the underlying cause in his case, though this was only a clinical observation. Just what he meant is not certain, though Rosenberg points out, that should he have implied an inhibition to ganglion cell growth ("Entwicklungshemmung") then this idea would be in keeping with Oppenheim's theory.

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[59] Münch. med. Woch., 1892.

[60] Berlin. klin. Woch., 1890. See also Lehrbuch, ii, 124, 125.

[61] Kunn, Beit. z. Augenheilkunde, Nos. 35 and 37 (cited by Zappert). See also Oppenheim, Lehrbuch, p. 1033.

[62] Heubner (Lehrbuch, ii, 124) speaks of being able at times to find a muscle wanting in other parts of the body having nothing to do with the bulbar nuclei (as the pectoralis major). The pectoralis as an isolated congenital defect, however, appears not to be uncommon. The literature has over a hundred cases. The combination with other muscle defects is rare. See especially Ziehen, Beziehungen zwichen angeborenen Muskaldefekten, infantilen Kernschwund, etc., Berl. klin. Woch., No. 34, August 24, 1908.

Of the dystrophies, we may say that the various types are usually (though not always) hereditary or familial—myatonia congenita being in but one instance familial[63] (Sorgente's two cases)—they are never congenital, they usually evidence severe atrophy, and generally affect the pelvic and lumbar musculature first, while the infantile type is earliest noticed in the face. Such distribution is not found in Oppenheim's disease. Electrically both conditions are not to be distinguished from each other, and in both a pseudohypotrophic form may occur because of adiposis, yet never has myatonia congenita resembled the typical picture of muscular dystrophy. Never has a case of muscular dystrophy improved as several myatonia cases have, nor have the patellar reflexes been seen to return in the former, as occasionally reported in the latter. Finally, and this is of greatest importance, the dystrophies are eminently progressive.

Much the same may be said of "early infantile spinal progressive muscle atrophy" (Werdnig and Hoffmann). This very uncommon disease (only about thirty cases have thus far been described) is exquisitely familial and hereditary (in one family six of fifteen children, in another three of six were affected; in one family in which three children were affected, eight brothers and sisters of the mother had died of the same disease). In this one fact it differs markedly from myatonia congenita, in all of which cases, with one exception (Sorgente's two cases in one family), there was no trace of any hereditary or familial tendency. Furthermore, as the majority of our myatonia cases are absolutely congenital; the cases of Werdnig, Hoffmann, and Bruns were all prenatal, beginning usually in the second half of the first year, in a few in the second year. Again all these cases were progressive and fatal, death occurring within two to four or five years. This progression showed itself in a spreading of the paralysis (beginning in the muscles of the pelvic girdle and thighs, gradually involving those of the trunk, shoulder girdle, neck, and, finally, the hands and feet). Parallel with the paralysis goes the atrophy, which becomes excessive. In no case of Oppenheim's disease has such progression been noticed, nor an outspoken atrophy. Nor was reaction of degeneration reported, though this was partially or completely present in a number of the Werdnig and Hoffmann type of cases. From this account it will be seen that though both diseases show a flaccid condition, with loss of tendon reflexes, a sparing of sensory, bladder, and rectal involvement, as well as of the muscular area innervated by cranial nerves, there is decided enough difference between them to diagnosticate separate clinical entities. The question, however, arises, Are there transition stages (Uebergangsformen) between the two conditions? Wimmer thought that his case was such,[64] and assumed that this disease constituted a condition in which a congenital resistentia minor obtained, which in several cases was temporary and reparable (Oppenheim's myatonia), in others progressive and lethal (Werdnig and Hoffmann's muscle-atrophy). And Rothmann[65] also maintained that the majority of myatonia cases are due to a lesion in the ganglion cells of the anterior horns occurring in fœtal life, which may vary in severity up to complete aplasia, and that between the myatonia of Oppenheim and the muscle atrophy of Hoffmann, all grades of transition cases occur, which are not to be sharply differentiated, though one may place the abating and improving myatonia cases apart from the increasing and pernicious spinal atrophies; and finally, that it is evident that transitions occur between the congenital spinal muscle atrophy with involvement of the bulbar nuclei and Möbius' infantile nuclei aplasia. Now, these seem to be interesting assumptions, though there appears to be too little in the data up to now available to substantiate them. The severe cases of myatonia did not gradually become so; the condition was seen in full development at or very soon after birth. Even in these severe cases no reaction of degeneration was present, nor was special atrophy seen; and Spiller's autopsy revealed nothing pathological in the nervous system of a pronouncedly affected case. This argues strongly against the spinal origin of the disease. On the other hand, the pathological findings reported by Baudouin speak for an arrest in the development, while the findings in the Werdnig and Hoffmann cases showed an outspoken atrophic degeneration of the entire spinomuscular neurone and terminating muscle.[66]

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[63] Because of this very fact, Collier and Wilson do not wish to count Sorgente's cases to those of myatonia congenita.

Interesting, too, is Bing's surmise that behind the myatonic condition may lie an inhibition not of muscle development, but of the tracts responsible for the regulation of muscle tonus—namely, the spinocerebellar tract. These tracts, however, were found in Baudouin's case to be intact. Berti, Cattaneo, and Baudouin thought that a congenital alteration in the internal secretion, probably hypofunction of the thyroid, was the cause of this disease. The thyroid gland in Baudouin's case did show extreme sclerosis. Is this, however, a chance concomitant occurrence, is it cause, or is it effect? There is no way of telling, though there is too much, as we have already seen, against the theory of thyroid hyposecretion to consider it as cause of the condition. And I may add to what I have already said under the differential diagnosis of myxœdema—and this applies to internal hyposecretion of thymus and adrenals—that one would expect in such disturbance an affection of the entire muscular system, and not of any special group of muscles (with sparing of other groups, as the distal muscles, those of bulbar innervation, etc.), as is the case in myatonia congenita.

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[64] The case was congenital, the atrophy and weakness progressive. The electrical reaction was much reduced, the galvanic tardy and with very little difference between CCC and ACC.

[65] Loc cit.

[66] Leyden-Goldscheider, Erkrankungen des Rückenmarkes, Nothnagel, Wien, 1904.

Upon the thought of a thymic syndrome (Smith), we can scarcely comment, as we know little or nothing about the thymus. It appears that a fibrosis of this gland is commonly associated with idiopathic infantile atrophies (Fortescue-Brickdale, Dudgeon[67]).

Very little can also be said of the microscopic muscle findings. These are not typical, but similar in large measure to those in the dystrophies and spinal atrophies. Furthermore, we know too little about fœtal and early infantile muscle conditions (there is really no literature to be found upon the subject) to determine to what extent these findings are to be considered pathological.

At the finale, therefore, we are left with our pathological problem unsolved. The few autopsy examinations which have been made lead us to believe the muscle changes to be secondary, not, however, to degenerative changes in the ganglion cells of the anterior horn, but to a retarded development of these or to a congenitally enfeebled or enervated condition—capable of developing, of improving, of regaining their "trophic virility." Whether in some cases these spinal cells are congenitally abiotrophic (the word is Gowers'), destined to degenerate and atrophy, it is as yet impossible to say. Of one thing we are, however, certain, and that is that this myatonia congenita of Oppenheim's is a new clinical entity, in its typical form a sharply defined syndrome differentiable from all the heretofore-known myopathies and spinal atrophies, constituting a congenital atonic or hypotonic, pseudoparalytic condition seen mostly in children within the first two years of infancy, associated with loss of tendon reflexes, without reaction of degeneration and atrophy, of proximal distribution, always affecting the lower yet often the upper extremities, trunk and neck beside, but not the muscles of bulbar innervation, and showing a tendency to spontaneous though slow improvement (hastened in many cases under massage and electrical treatment).

[67] Cited by Carey Coombs.

[68] Bull. de la Société de pédiatrie de Paris, Févrer-Mars, 1899.

[69] Lévi-Singue, L'atonie Musculaire Congénitale, Gazette des Hôpitaux, No. 15, Fevrier, 1909.